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McCune-Albright Syndrome Progressing With Severe Fibrous Dysplasia

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We present the case of an 11-year-old girl with McCune-Albright syndrome associated with severe fibrous dysplasia. In addition to bone lesions, she has apparent manifestations of precocious puberty. In examination, a mass at the mentum spreading to mandibular corpus bilaterally was seen. This mass has affected the mandibular teeth. It was nearly 20 × 20 × 15 cm in size and had local necrotic regions on it. The lower lip was expanded too much by the mass. Another mass filled the left maxillary sinus, expanding the left zygomatic region outwardly and closing the left nasal fossa completely. A rectangular skull shape was related to the involvement of cranial bones. After stainless steel, custom-made mandibular prosthesis was prepared, the patient underwent surgery. A partial mandibulectomy was performed, and resulting mandibular bone defect was reconstructed by steel prosthesis. Craniofacial involvement occurs in 100% of disseminated cases. In the dental literature, mandibular involvement was found in 20% of cases. However, in studied literature, we did not find a dramatic mandibular lesion as severe as that presented here.

Key Words: McCune-Albright syndrome, bone defect

The classic form of McCune-Albright syndrome, first described in 1930, is characterized by brown, patchy pigmentations on skin, polyostatic fibrous dysplasia, and precocious puberty.[1,2] The syndrome has also been called osteitis fibrosa cystica, osteodystrophia fibrosa, and osteofibroma, until it was named McCune-Albright syndrome. More than 100 cases have been reported, and the male–female ratio is 3:2.[3]

In general, skin lesions are evident in early infancy and are of the café au lait type. As the patient ages, these pigmentations may become darker brown. A direct proportion between the size of the pigmentations and the severity of bone involvement and endocrinological dysfunctions has been reported.[4,5] Endocrine disorders are mainly represented as sexual precocity, which is particularly evident in girls. Laboratory findings are usually within normal limits except for an occasional rise in alkaline phosphatase and estrogen levels.[6]

The origin of the sexual precocity in McCune-Albright syndrome is still unclear, just as are other components of the disease. It is reported that development of precocious puberty as an ovarian hyperfunction may be dependent or independent of gonadotropins.[7]

The character of fibrous dysplasia in this syndrome is of polyostotic type.[4,8,9] The proximal aspects of the femur and pelvis are characteristically involved. A bowing like a hockey stick may be produced, resulting in leg length discrepancy.[5,10] Polyostatic fibrous dysplasia shows a 20% predilection for the mandible. In patients with moderate skeletal involvement, about 50% of lesions are craniofacial. With extensive skeletal involvement, the incidence of fibrous dysplasia in the craniofacial region approaches 100%.[11]

Other endocrine dysfunctions such as hyperthyroidism, hyperparathyroidism, Cushing's syndrome, and hyperprolactinemia can be seen in McCune-Albright syndrome. The reasons of endocrine overproduction are not understood.[7,12]

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Fig 1 General appearance of the patient (lateral view).

Patient History

An 11-year-old girl was admitted for intraoral painless swelling of the mandible, which is expanding throughout mouth (Figs 1 and 2).

There was no reported consanguinity between parents, and the patient's two sisters were healthy. Café au lait pigmentations have been present since birth. The patient underwent surgery five times because of fractures of femur after minor trauma. Telarche and pubarche began at 3 years of age and menstruation at 5 years. McCune-Albright syndrome was diagnosed. The mass at her chin was seen and began to grow at the age of 4 years. A resection was performed by electrocautery to the mass when the patient was 8 years old. It had begun to grow in the postoperative period and reached its ultimate size in 3 years.

In her examination, a mass at the mentum, spreading to the mandibular corpus bilaterally and affecting the mandibular integrity and all the teeth localizations at the affected region, was revealed. It was nearly 20 × 20 × 15 cm in size and had local necrotic regions on it. The lower lip was expanded to a great degree by the mass (see Figs 1 through 3). Another mass was noted that filled the left maxillary sinus, expanding the left zygomatic region outwardly and closing the left nasal fossa completely (Fig 2). A rectangular skull shape was determined to be related to cranial bone involvement. There were large patches of café au lait spots on her torso and face. Because of the involvement of both of the femurs and the performed operations, there were shortness and severe deformity in her lower extremities. Precocious puberty was thought because of her axillary and pubic hair growth, breast development, and menstruation.

Whole values of complete blood count and blood chemistry were in normal range except increased alkaline phosphatase, which was 2,521 U/L (normal, 98–279). All hormonal investigations were found to be normal. However, reproductive hormones were in concert with precocious puberty.

A craniofacial tomographic scan revealed fibrous dysplasia affecting the cranial and facial bones, and a considerable thickening in calvarian bones was

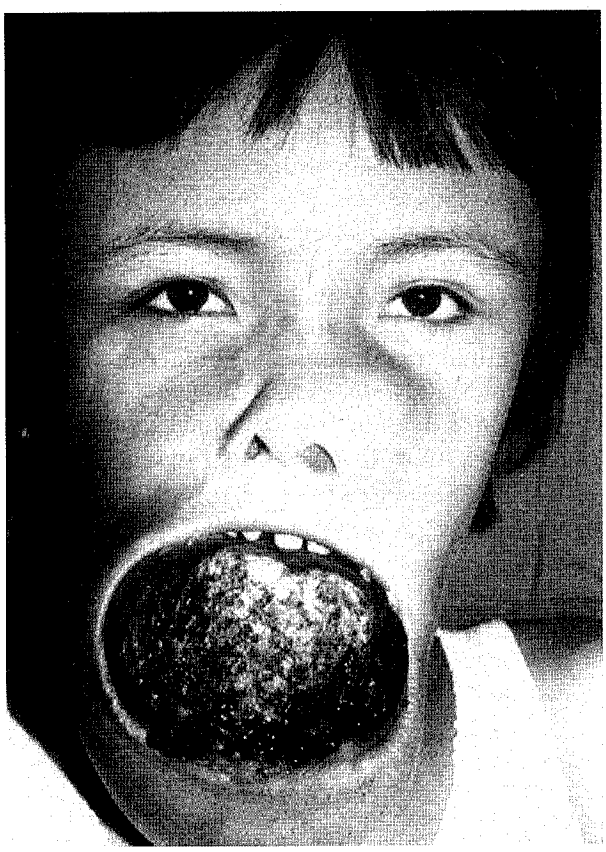

Fig 2 Anterior aspect of mandibular mass.

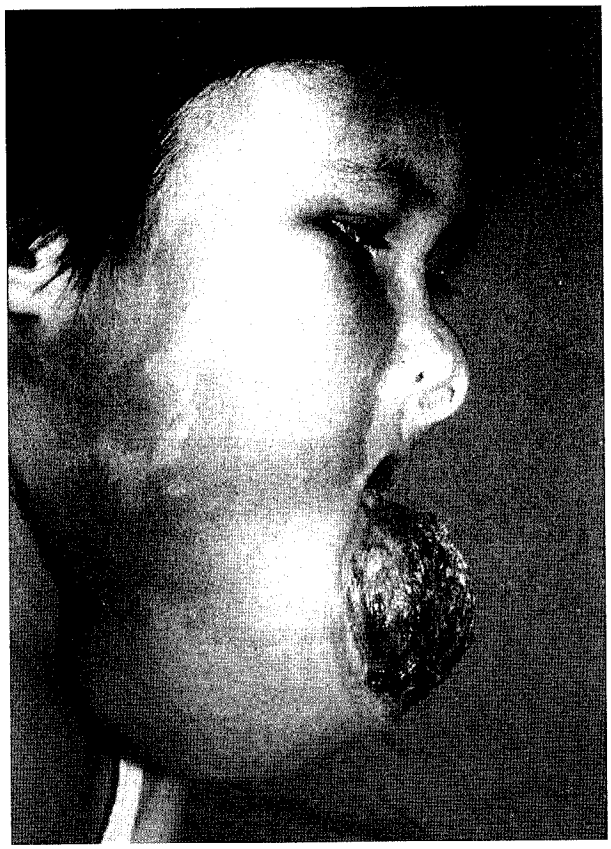

Fig 3 Lateral aspect of mandibular mass.

found (Fig 4). The patient underwent surgery after the completion of routine analysis. The expanded lower lip was opened at the midline. We reached to the tumoral mass and mandible with mucosal incisions through the lower vestibular fold. Although the entire mandible was attacked by fibrous dysplasia, a partial mandibulectomy and tumoral mass excision were performed. Afterward, mandibular parts were carved in both sides and the tips of a custom-made prothesis were placed and was fixed with bone cement.

The lower lip was joined at the midline, and its expanded parts were removed at the upper area. A Weber-Ferguson-Longwire incision was made on the left side, and the mass, which covered the left maxillary sinus was excised with chisels. The operation was completed by suturing all the incisions. No surgical complication developed postoperatively (Fig 5).

Even though pre- and postoperative routine analyses were in normal range, the postoperative blood glucose level was 294 mg%. Required insulin management and diet were implemented. On postoperative day 3, blood glucose level was normal.

Discussion

Pediatricians and endocrinologists are concerned about McCune-Albright syndrome. Because nothing about its cause and pathogenesis is certain, patient follow-up and treatment are crucial. Of the three components of this syndrome—multifocal fibrous dysplasia in bones, widespread café au lait spots, and precocious puberty—the latter is the most important.

Pathologic fractures, especially in the femur, caused our patient to crawl on all fours. Because craniofacial involvement occurs in 100% of disseminated cases, plastic surgeons are often consulted. In such involvements, multiple pathological cranial base fractures, deafness, optic atrophy, and neuralgia caused by pressure to cranial nerves were reported.[12] Although it is rarely reported in dental literature,[6] in 20% of our cases, mandibular involvement was found. However, in studied literature, these mandibular lesions were not found to be as dramatic or severe as in our patient. In fibrous dysplasia associated with McCune-Albright's syndrome, a malignant degeneration such as osteogenic sarcoma was re-

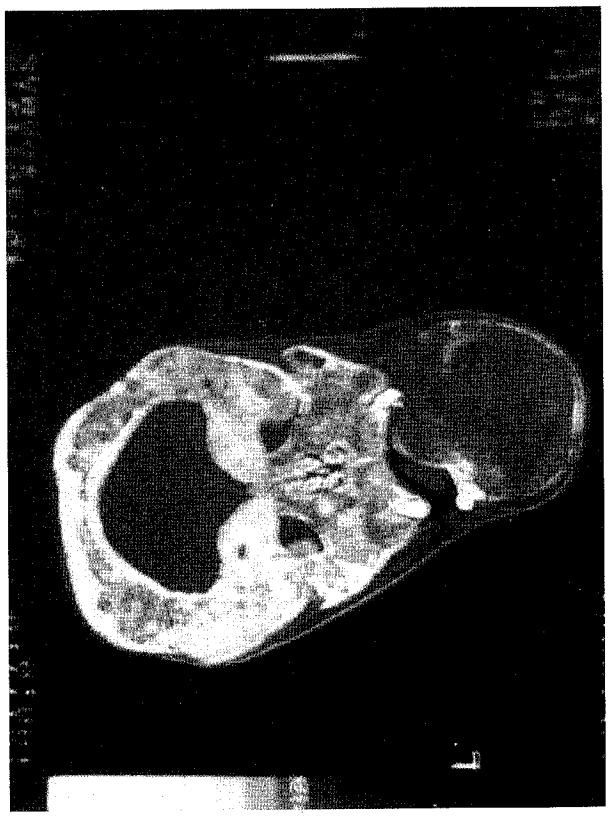

Fig 4 Tomographic view of the lesion.

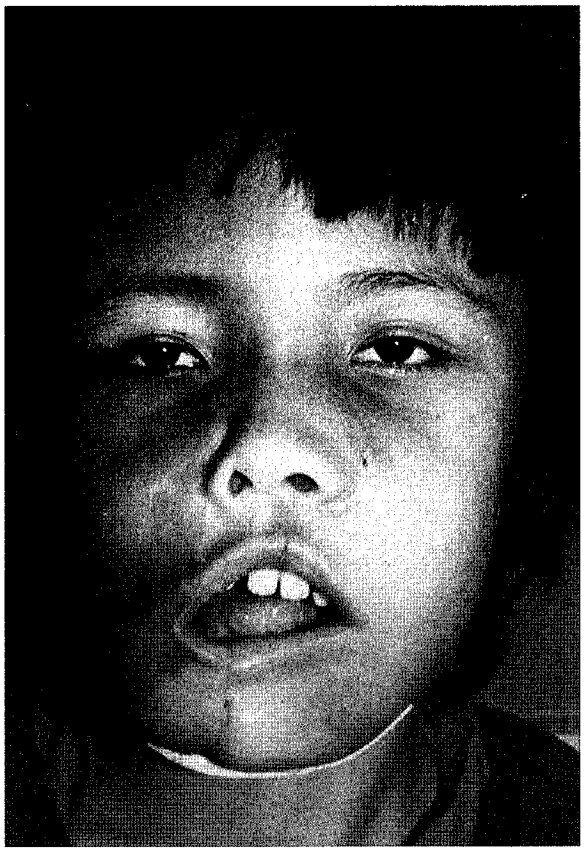

Fig 5 Postoperative appearance of the patient.

ported.[13] There was no malignancy in this case. Endocrinological disorders, such as Cushing's disease, goiter, hyperparathyroidism, acromegaly, hypothalamic diabetes mellitus, hyperthyroidism, and hyperprolactinemia may be associated with this syndrome.[6,7,12]

Hyperparathyroidism, histiocytosis, multiple myeloma, Paget's disease, juvenile Paget's disease, neurofibromatosis, and cherubism should be included in the differential diagnosis of bone lesions of McCune-Albright's syndrome.[14]

Some endocrinologic disorders such as thyroid storm have been reported in surgical patients with McCune-Albright syndrome[15]; however, no problem has occurred in our patient other than hyperglycemia, which continued in first 2 postoperative days.

Our patient is still under observation by the Department of Pediatric Endocrinology to detect new genetic findings in light of the latest literature and to manage the therapeutic procedures.

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